L Number	Hits	Search Text	DB	Time stamp
1	2491	stoffel	USPAT; US-PGPUB	2002/09/13 07:43
2	1506	haplotyp$4	USPAT; US-PGPUB	2002/09/13 07:43
3	15012	allel$4	USPAT; US-PGPUB	2002/09/13 07:43
4	136	pasa	USPAT; US-PGPUB	2002/09/13 07:43
5	469155	arms	USPAT; US-PGPUB	2002/09/13 07:44
6	22	stoffel same arms	USPAT; US-PGPUB	2002/09/13 07:46
7	0	stoffel same pasa	USPAT; US-PGPUB	2002/09/13 07:45
8	2	stoffel same haplotyp$4 same allel$4	USPAT; US-PGPUB	2002/09/13 07:46
9	4	stoffel same allel$4	USPAT; US-PGPUB	2002/09/13 07:47

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=> file medline biosis caplus
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=> s (double (w) arms or PASA)
L1          357 (DOUBLE (W) ARMS OR PASA)

=> s l1 and RFLP
L2           16 L1 AND RFLP

=> s l1 and stoffel
L3            0 L1 AND STOFFEL

=> s l1 and exonuclease#
L4            0 L1 AND EXONUCLEASE#

=> s l1 and exo?
L5           19 L1 AND EXO?

=> dup rem l5
PROCESSING COMPLETED FOR L5
L6           10 DUP REM L5 (9 DUPLICATES REMOVED)

=> d 1-10 ri
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L6   ANSWER 1 OF 10  BIOSIS  COPYRIGHT 2002 BIOLOGICAL ABSTRACTS INC.
TI   Nucleic acid amplification with direct sequencing.

L6   ANSWER 2 OF 10  BIOSIS  COPYRIGHT 2002 BIOLOGICAL ABSTRACTS INC.
TI   Nucleic acid amplification with direct sequencing.

L6   ANSWER 3 OF 10     MEDLINE                          DUPLICATE 1
TI   Characterization and screening of a point mutation in LDL receptor gene
     found in southern Italy (FHAvellino).

L6   ANSWER 4 OF 10     MEDLINE                          DUPLICATE 2
TI   Dopamine receptor gene polymorphisms in Guangzhou Hans.

L6   ANSWER 5 OF 10     MEDLINE                          DUPLICATE 3
TI   Bi-PASA genotyping of a new polymorphism in the APOB gene shows
     no evidence for an association with fatness in pigs.

L6   ANSWER 6 OF 10  CAPLUS  COPYRIGHT 2002 ACS
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TI Simple detection of a point mutation in LDL receptor gene causing familial hypercholesterolemia in southern Italy by allele-specific polymerase chain reaction

L6 ANSWER 7 OF 10 MEDLINE DUPLICATE 4
TI Identification of a leucine-to-proline mutation in the keratin 5 gene in a family with the generalized Kobner type of epidermolysis bullosa simplex.

L6 ANSWER 8 OF 10 MEDLINE DUPLICATE 5
TI Two-tiered DNA-based diagnosis of transthyretin amyloidosis reveals two novel point mutations.

L6 ANSWER 9 OF 10 MEDLINE DUPLICATE 6
TI [The effect of tuberculous intoxication and tuberculostatic preparations on pancreatic excretory function].
Vliianie tuberkuleznoi intoksikatsii i tuberkulostaticheskikh preparatov na ekskretornuiu funktsiiu podzheludochnoi zhelezy.

L6 ANSWER 10 OF 10 MEDLINE
TI Transforming gene in the preleukemic state.

=> d 5 bib ab

L6 ANSWER 5 OF 10 MEDLINE DUPLICATE 3
AN 1999159192 MEDLINE
DN 99159192 PubMed ID: 10050285
TI Bi-**PASA** genotyping of a new polymorphism in the APOB gene shows no evidence for an association with fatness in pigs.
AU Jiang Z H; Gibson J P
CS Department of Animal and Poultry Science, University of Guelph, Ontario, Canada.
SO ANIMAL GENETICS, (1999 Feb) 30 (1) 54-6.
Journal code: 8605704. ISSN: 0268-9146.
CY ENGLAND: United Kingdom
DT Journal; Article; (JOURNAL ARTICLE)
LA English
FS Priority Journals
EM 199903
ED Entered STN: 19990402
Last Updated on STN: 19990402
Entered Medline: 19990324
AB Sequence analysis of PCR products of a 343-bp fragment from **exon** 29 of the porcine APOB gene of four Erhualian and four Landrace pigs revealed a missense G/C substitution at position of 6117 in this gene. Two allele-specific primers were designed to genotype this polymorphism using the Bi-**PASA** technique. Genotyping of 146 animals from Erhualian, Hampshire, Large White, Landrace and Duroc breeds revealed large breed differences in allele frequency. No association with fatness was observed within each of the four European breeds, where animals had been selected to be those with the highest and those with the lowest backfat depth at 100 kg liveweight from a large carcass dissection project.

=> d 5 all

L6 ANSWER 5 OF 10 MEDLINE DUPLICATE 3
AN 1999159192 MEDLINE
DN 99159192 PubMed ID: 10050285
TI Bi-**PASA** genotyping of a new polymorphism in the APOB gene shows no evidence for an association with fatness in pigs.
AU Jiang Z H; Gibson J P

CS Department of Animal and Poultry Science, University of Guelph, Ontario, Canada.
SO ANIMAL GENETICS, (1999 Feb) 30 (1) 54-6.
Journal code: 8605704. ISSN: 0268-9146.
CY ENGLAND: United Kingdom
DT Journal; Article; (JOURNAL ARTICLE)
LA English
FS Priority Journals
EM 199903
ED Entered STN: 19990402
Last Updated on STN: 19990402
Entered Medline: 19990324
AB Sequence analysis of PCR products of a 343-bp fragment from **exon** 29 of the porcine APOB gene of four Erhualian and four Landrace pigs revealed a missense G/C substitution at position of 6117 in this gene. Two allele-specific primers were designed to genotype this polymorphism using the Bi-**PASA** technique. Genotyping of 146 animals from Erhualian, Hampshire, Large White, Landrace and Duroc breeds revealed large breed differences in allele frequency. No association with fatness was observed within each of the four European breeds, where animals had been selected to be those with the highest and those with the lowest backfat depth at 100 kg liveweight from a large carcass dissection project.
CT Check Tags: Animal
*Apolipoproteins B: GE, genetics
Gene Frequency
Genotype
Mutation, Missense
Obesity: GE, genetics
*Obesity: VE, veterinary
Polymerase Chain Reaction: MT, methods
Polymerase Chain Reaction: VE, veterinary
*Polymorphism (Genetics)
*Swine: GE, genetics
CN 0 (Apolipoproteins B)

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L6 ANSWER 5 OF 10 MEDLINE DUPLICATE 3
TI Bi-**PASA** genotyping of a new polymorphism in the APOB gene shows no evidence for an association with fatness in pigs.
AB Sequence analysis of PCR products of a 343-bp fragment from **exon** 29 of the porcine APOB gene of four Erhualian and four Landrace pigs revealed a missense G/C substitution at position of 6117 in this gene. Two allele-specific primers were designed to genotype this polymorphism using the Bi-**PASA** technique. Genotyping of 146 animals from Erhualian, Hampshire, Large White, Landrace and Duroc breeds revealed large breed differences in allele. . .

=> d his

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FILE 'MEDLINE, BIOSIS, CAPLUS' ENTERED AT 07:34:54 ON 13 SEP 2002
L1 357 S (DOUBLE (W) ARMS OR PASA)

L2 16 S L1 AND RFLP
L3 0 S L1 AND STOFFEL
L4 0 S L1 AND EXONUCLEASE#
L5 19 S L1 AND EXO?
L6 10 DUP REM L5 (9 DUPLICATES REMOVED)

=> d 1-10 bib ab

L6 ANSWER 1 OF 10 BIOSIS COPYRIGHT 2002 BIOLOGICAL ABSTRACTS INC.
AN 2002:279713 BIOSIS
DN PREV200200279713
TI Nucleic acid amplification with direct sequencing.
AU Sommer, Steven Seev (1)
CS (1) Rochester, MN USA
ASSIGNEE: Sommer; Steve S., Duarte, CA, USA
PI US 6361949 March 26, 2002
SO Official Gazette of the United States Patent and Trademark Office Patents, (Mar. 26, 2002) Vol. 1256, No. 4, pp. No Pagination. http://www.uspto.gov/web/menu/patdata.html. e-file. ISSN: 0098-1133.
DT Patent
LA English
AB This invention provides methods of amplifying a sequence of interest present within a nucleic acid molecule. In addition, this invention provides a method of determining the nucleotide sequence of a sequence of interest present within a nucleic acid molecule (e.g. GAWTS and RAWTS) which can be used to sequence tissue specific genes (e.g. tsRAWTS) and genes across species (e.g. zooRAWTS). In addition, this invention provides a method of synthesizing a polypeptide encoded for by a nucleic acid molecule (RAWIT). Further, the subject invention provides a method of determining an internal nucleotide sequence present within a nucleic acid molecule, and a method of determining a terminal nucleotide sequence present within a nucleic acid molecule (e.g. PLATS). Also provided for is a method of determining the nucleotide sequence of sequences present within a nucleic acid molecule which are adjacent to areas of known sequence (e.g ASWATS) and a method of determining the nucleotide sequence of sequences present within a nucleic acid molecule and a method of detecting point mutation or polymorphism (e.g. **PASA**) which can be used in low cost methods of carrier testing and prenatal diagnosis. Lastly, this invention provides methods for determining the **exonic** nucleotide sequence of a gene as well as methods of detecting genomic mutations.

L6 ANSWER 2 OF 10 BIOSIS COPYRIGHT 2002 BIOLOGICAL ABSTRACTS INC.
AN 2000:369546 BIOSIS
DN PREV200000369546
TI Nucleic acid amplification with direct sequencing.
AU Sommer, Steven S. (1)
CS (1) 2317 Viking Dr., Northwest Rochester, MN, 55901 USA
PI US 6027913 February 22, 2000
SO Official Gazette of the United States Patent and Trademark Office Patents, (Feb. 22, 2000) Vol. 1231, No. 4, pp. No pagination. e-file. ISSN: 0098-1133.
DT Patent
LA English
AB This invention provides methods of amplifying a sequence of interest present within a nucleic acid molecule. In addition, this invention provides a method of determining the nucleotide sequence of a sequence of interest present within a nucleic acid molecule (e.g. GAWTS and RAWTS) which can be used to sequence tissue specific genes (e.g. tsRAWTS) and genes accross species (e.g. zooRAWTS). In addition, this invention provides a method of synthesizing a polypeptide encoded for by a nucleic

acid molecule (RAWIT). Further, the subject invention provides a method of determining an internal nucleotide sequence present within a nucleic acid molecule, and a method of determining a terminal nucleotide sequence present within a nucleic acid molecule (e.g. PLATS). Also provided for is a method of determining the nucleotide sequence of sequences present within a nucleic acid molecule which are adjacent to areas of known sequence (e.g ASWATS) and a method of determining the nucleotide sequence of sequences present within a nucleic acid molecule and a method of detecting point mutation or polymorphism (e.g. **PASA**) which can be used in low cost methods of carrier testing and prenatal diagnosis. Lastly, this invention provides methods for determining the **exonic** nucleotide sequence of a gene as well as methods of detecting genomic mutations.

L6 ANSWER 3 OF 10 MEDLINE DUPLICATE 1
AN 2001272694 MEDLINE
DN 21261224 PubMed ID: 11367925
TI Characterization and screening of a point mutation in LDL receptor gene found in southern Italy (FHAvellino).
AU Cantafora A; Blotta I; Mercuri E; Cortese C; Motti C; Rampa P; Calandra S; Bertolini S
CS Laboratorio di Metabolismo e Biochimica Patologica, Istituto Superiore di Sanita, Rome, Italy.
SO ANNALI DELL ISTITUTO SUPERIORE DI SANITA, (2000) 36 (4) 459-64.
Journal code: 7502520. ISSN: 0021-2571.
CY Italy
DT Journal; Article; (JOURNAL ARTICLE)
LA English
FS Priority Journals
EM 200109
ED Entered STN: 20010910
Last Updated on STN: 20010910
Entered Medline: 20010906
AB The finding that the missense mutation C331W in the **exon** 7 of LDL-receptor gene, previously reported to occur in Holland and Belgium, caused the homozygote familial hypercholesterolemia (FH) in an individual from the district of Avellino induced us to search the mutation in a large area of region Campania. This was made with simple screening methods developed by ourselves and based on either the recognition of a primer-induced Fok I restriction site in the mutant allele or the PCR allele-specific amplification (**PASA**) of mutant allele. They were applied to a total of 144 unrelated cases recruited from where the mutation was more likely to occur. We failed to reveal any new case of C331W mutation that is indeed not common within the area of this screening, at spite of having been found in different countries.

L6 ANSWER 4 OF 10 MEDLINE DUPLICATE 2
AN 2001026391 MEDLINE
DN 20482098 PubMed ID: 11024217
TI Dopamine receptor gene polymorphisms in Guangzhou Hans.
AU Wang J; Liu Z; Chen B; Li J; Pan Y; Lu X; Zhu Y
CS Department of Neurology, the First Affiliated Hospital, Sun Yat-sen University of Medical Sciences, Guangzhou, 510080 P.R.China..
jian-w@163.net
SO CHUNG-HUA I HSUEH I CHUAN HSUEH TSA CHIH, (2000 Oct) 17 (5) 348-51.
Journal code: 9425197. ISSN: 1003-9406.
CY China
DT Journal; Article; (JOURNAL ARTICLE)
LA Chinese
FS Priority Journals
EM 200011
ED Entered STN: 20010322

Last Updated on STN: 20010322
Entered Medline: 20001116

AB OBJECTIVE: To determine the distribution of dopamine D2, D3, and D5 receptor(DRD2, DRD3, DRD5) gene polymorphisms in Guangzhou Hans. METHODS: A total of 141 healthy Guangzhou Hans were studied by the use of polymerase chain reaction-restriction fragment length polymorphism (PCR-RFLP) and PCR-amplification of specific allele (**PASA**) techniques. Current results were compared with the data on other ethnic groups. RESULTS: Within the 141 individuals tested, the frequencies of the A1(Taq I-) and A2(Taq I+) alleles of the TaqI A mutation site in the 3'non-coding region of the DRD2 gene were found to be 48% and 52%, respectively. The observed frequencies of the A1A1, A1A2 and A2A2 genotypes were found to be 17%, 52% and 31%, respectively, which met Hardy-Weinberg equilibrium. The frequencies of the A1 and A2 alleles of the Bal I mutation site in the **exon** 1 of DRD3 gene were found to be 73% and 27%, respectively. The observed frequencies of the A1A1, A1A2 and A2A2 genotypes were found to be 53%, 39% and 8%, respectively, which met Hardy-Weinberg equilibrium. The frequencies of the 1 and 2 alleles of the Msp I mutation site in the intron 5 of DRD3 gene were found to be 62.5% and 37.5%, respectively. The observed frequencies of the 1-1, 1-2 and 2-2 genotypes were found to be 35%, 55% and 10%, respectively, which met Hardy-Weinberg equilibrium. No linkage disequilibrium was observed between the Bal I and Msp I polymorphism in Guangzhou Hans(chi(2)=0165, P>0.05). The frequencies of the T and C alleles of the T978C mutation site of DRD5 gene were found to be 51% and 49%, respectively. The observed frequencies of the T/T, T/C and C/C genotypes were found to be 23.6%, 54.6% and 21.8%, respectively, which met Hardy-Weinberg equilibrium. CONCLUSION: The polymorphisms of DRD2, DRD3, DRD5 gene in Guangzhou Hans were high and different from those in other populations.

L6 ANSWER 5 OF 10 MEDLINE DUPLICATE 3
AN 1999159192 MEDLINE
DN 99159192 PubMed ID: 10050285
TI Bi-**PASA** genotyping of a new polymorphism in the APOB gene shows no evidence for an association with fatness in pigs.
AU Jiang Z H; Gibson J P
CS Department of Animal and Poultry Science, University of Guelph, Ontario, Canada.
SO ANIMAL GENETICS, (1999 Feb) 30 (1) 54-6.
Journal code: 8605704. ISSN: 0268-9146.
CY ENGLAND: United Kingdom
DT Journal; Article; (JOURNAL ARTICLE)
LA English
FS Priority Journals
EM 199903
ED Entered STN: 19990402
Last Updated on STN: 19990402
Entered Medline: 19990324

AB Sequence analysis of PCR products of a 343-bp fragment from **exon** 29 of the porcine APOB gene of four Erhualian and four Landrace pigs revealed a missense G/C substitution at position of 6117 in this gene. Two allele-specific primers were designed to genotype this polymorphism using the Bi-**PASA** technique. Genotyping of 146 animals from Erhualian, Hampshire, Large White, Landrace and Duroc breeds revealed large breed differences in allele frequency. No association with fatness was observed within each of the four European breeds, where animals had been selected to be those with the highest and those with the lowest backfat depth at 100 kg liveweight from a large carcass dissection project.

L6 ANSWER 6 OF 10 CAPLUS COPYRIGHT 2002 ACS
AN 1998:312080 CAPLUS
DN 129:77154

TI Simple detection of a point mutation in LDL receptor gene causing familial hypercholesterolemia in southern Italy by allele-specific polymerase chain reaction

AU Cantafora, Alfredo; Blotta, Ida; Mercuri, Elisabetta; Calandra, Sebastiano; Bertolini, Stefano

CS Laboratorio di Metabolismo e Biochimica Patologica, Istituto Superiore di Sanita, Rome, 00161, Italy

SO Journal of Lipid Research (1998), 39(5), 1101-1105
CODEN: JLPRAW; ISSN: 0022-2275

PB Lipid Research, Inc.

DT Journal

LA English

AB Polymerase chain reaction (PCR) amplification of specific alleles allowed the rapid detection of a point mutation (missense Gly528 .fwdarw. Asp) in **exon** 11 of the low d. lipoprotein receptor gene which was otherwise not detectable by **exon** amplification and enzymic digestion as it does not modify the normal restriction pattern. The mutant allele, designated as FH-Palermo-1 from the origin of the first carrier family identified, gave a specific PCR product of 109 bp clearly distinct from the product of 168 bp obtained from other alleles with a nonspecific couple of primers. This method allowed us to distinguish one pos. sample mixed with up to 11 parts of normal DNA. Furthermore, the specific amplification product was characterized by a Bsm I restriction site not present in nonspecific products.

L6 ANSWER 7 OF 10 MEDLINE DUPLICATE 4

AN 93306317 MEDLINE

DN 93306317 PubMed ID: 7686424

TI Identification of a leucine-to-proline mutation in the keratin 5 gene in a family with the generalized Kobner type of epidermolysis bullosa simplex.

AU Dong W; Ryynanen M; Uitto J

CS Department of Dermatology, Jefferson Medical College, Thomas Jefferson University, Philadelphia, Pennsylvania 19107.

NC PO1-AR38923 (NIAMS)
T32-AR07561 (NIAMS)

SO HUMAN MUTATION, (1993) 2 (2) 94-102.
Journal code: 9215429. ISSN: 1059-7794.

CY United States

DT Journal; Article; (JOURNAL ARTICLE)

LA English

FS Priority Journals

EM 199308

ED Entered STN: 19930813
Last Updated on STN: 19960129
Entered Medline: 19930805

AB We have previously reported linkage of a large Finnish family with the generalized (Kobner) type of epidermolysis bullosa simplex to chromosome 12q in the region containing the type II keratin gene cluster (Ryynanen et al., Am J Human Genet 49:978-984, 1991). In this study, we examined the possibility that keratin 5, the type II keratin expressed in the basal keratinocytes, harbors the mutation in this family. Nucleotide sequencing revealed a T-to-C transition within **exon** 7 of the keratin 5 gene in the affected individuals of the family, while the unaffected individuals showed no evidence of C. The presence of the T-to-C transition in the affected individuals was confirmed by restriction enzyme digestion analysis with NciI endonuclease, as well as with PCR amplification of specific alleles (**PASA**) analysis. The **PASA** analysis also indicated that the mutated allele was not found among the 100 alleles tested within the general Finnish population indicating that the mutated allele is not a common polymorphism. Furthermore, the mutated allele was not present in nine individuals representing three different EBS families of Finnish origin. The T-to-C transition at the nucleotide level resulted

in substitution of a leucine by a proline at the amino acid level, and the substitution affected a leucine residue which was invariant among eight different human keratins in a highly conserved segment at the carboxy-terminal region of the keratin 5 polypeptide.(ABSTRACT TRUNCATED AT 250 WORDS)

L6 ANSWER 8 OF 10 MEDLINE DUPLICATE 5
AN 91261170 MEDLINE
DN 91261170 PubMed ID: 2046936
TI Two-tiered DNA-based diagnosis of transthyretin amyloidosis reveals two novel point mutations.
AU Ii S; Minnerath S; Ii K; Dyck P J; Sommer S S
CS Department of Neurology, Mayo Clinic, Rochester, MN 55905.
SO NEUROLOGY, (1991 Jun) 41 (6) 893-8.
Journal code: 0401060. ISSN: 0028-3878.
CY United States
DT Journal; Article; (JOURNAL ARTICLE)
LA English
FS Abridged Index Medicus Journals; Priority Journals
EM 199107
ED Entered STN: 19910802
Last Updated on STN: 19910802
Entered Medline: 19910716
AB We analyzed 11 consecutive unrelated cases of polyneuropathy due to transthyretin amyloidosis. Direct sequencing of the promoter region, **exons**, and splice junctions revealed that each patient was heterozygous for a mutation: six patients had valine 30 substituted by methionine (V30----M; Portuguese-Japanese type), one had threonine 60 substituted by alanine (T60----A; Appalachian type), and two had serine 77 substituted by tyrosine (S77----Y; Illinois type). In addition, two patients had previously undescribed mutation: phenylalanine 33 substituted by leucine (F33----L) and phenylalanine 64 substituted by leucine (F64----L). From present information, the probands of these novel mutations do not exhibit any pathology that clearly distinguishes them from individuals with the other mutations. The mutations extend the range of mutations associated with amyloidotic polyneuropathy. In our 11 patients, the different mutations did not seem to correlate with distinct clinical phenotypes. We developed **PASA** assays (PCR amplification of specific alleles) for each of the five mutations. **PASA** can be used by any diagnostic laboratory that can perform PCR to rapidly detect any of the known mutations. The minority of samples with an undescribed mutation can be sent to a specialty laboratory for delineation of the mutation by direct genomic sequencing. The presently described combination of methods may have widespread utility in the diagnosis of genetic disease.

L6 ANSWER 9 OF 10 MEDLINE DUPLICATE 6
AN 90192695 MEDLINE
DN 90192695 PubMed ID: 2483460
TI [The effect of tuberculous intoxication and tuberculostatic preparations on pancreatic excretory function].
Vliianie tuberkuleznoi intoksikatsii i tuberkulostaticheskikh preparatov na ekskretornuiu funktsiiu podzheludochnoi zhelezy.
AU Plemiannikova G I
SO PROBLEMY TUBERKULEZA, (1989) (12) 55-8.
Journal code: 0414141. ISSN: 0032-9533.
CY USSR
DT Journal; Article; (JOURNAL ARTICLE)
LA Russian
FS Priority Journals
EM 199004
ED Entered STN: 19900601

Last Updated on STN: 19960129
Entered Medline: 19900426

AB The excretory function of the pancreas was examined intraduodenally in 78 patients with various forms of pulmonary tuberculosis. There was a decrease in its **exocrine** function. It was shown that the medium and conditions in the duodenum cavity affected the enzyme activity in it. The intraduodenal amylolytic activity was lowered and no effects of the antituberculous drugs on it was observed. The intraduodenal proteolytic activity was increased. which was due to tuberculous intoxication and the pharmacological action of the tuberculostatic drugs, especially **PASA**. The causes of the adverse dyspeptic reactions in the patients subjected to the specific therapy including **PASA** were revealed.

L6 ANSWER 10 OF 10 MEDLINE
AN 87269684 MEDLINE
DN 87269684 PubMed ID: 3606144
TI Transforming gene in the preleukemic state.
AU Takaku F; Hirai H; Nishida J
SO GAN TO KAGAKU RYOHO [JAPANESE JOURNAL OF CANCER AND CHEMOTHERAPY], (1987 Jun) 14 (6 Pt 2) 2170-5.
Journal code: 7810034. ISSN: 0385-0684.
CY Japan
DT Journal; Article; (JOURNAL ARTICLE)
LA Japanese
FS Priority Journals
EM 198707
ED Entered STN: 19900305
Last Updated on STN: 19900305
Entered Medline: 19870727

AB The presence of a transforming gene in the DNA of bone marrow cells from patients with myelodysplastic syndrome (MDS) was studied using an in vivo selection assay in which NIH 3T3 cells transfected with human DNA were injected into nude mice in order to observe the growth of the tumor. The transforming gene was present in 12 out of 18 cases. The Alu sequence was demonstrated in the tumor grown after injection of transfected NIH 3T3 cells from 10 patients. Among these 10 Alu sequences, the human N-ras oncogene was present in 3 cases. Analysis of nucleotide sequences of the **exons** of human N-ras oncogenes cloned from the tumors revealed a single point mutation of the codon encoding the 13th amino acid of **exon** 1 from guanine to cytosine in all 3 cases of MDS. A one-year follow-up study of these MDS cases showed that in the patients positive for the transforming gene, the disease state progressed from PARA, **PASA** to RAEB or from RAEB to acute leukemia in 6 out of 7 cases, while in the 6 negative patients, no change was observed in their disease states. It was considered that the mutation of the N-ras gene at the 13th amino acid codon of **exon** 1 was fairly specific to MDS and that presence of the transforming gene may be used for predicting the progress of the disease.

=> d his

(FILE 'HOME' ENTERED AT 07:34:20 ON 13 SEP 2002)

FILE 'MEDLINE, BIOSIS, CAPLUS' ENTERED AT 07:34:54 ON 13 SEP 2002
L1 357 S (DOUBLE (W) ARMS OR PASA)
L2 16 S L1 AND RFLP
L3 0 S L1 AND STOFFEL
L4 0 S L1 AND EXONUCLEASE#
L5 19 S L1 AND EXO?
L6 10 DUP REM L5 (9 DUPLICATES REMOVED)

=> dup rem l2
PROCESSING COMPLETED FOR L2
L7 10 DUP REM L2 (6 DUPLICATES REMOVED)

=> d 1-10 bib ab

L7 ANSWER 1 OF 10 MEDLINE DUPLICATE 1
AN 2001026391 MEDLINE
DN 20482098 PubMed ID: 11024217
TI Dopamine receptor gene polymorphisms in Guangzhou Hans.
AU Wang J; Liu Z; Chen B; Li J; Pan Y; Lu X; Zhu Y
CS Department of Neurology, the First Affiliated Hospital, Sun Yat-sen University of Medical Sciences, Guangzhou, 510080 P.R.China.. jian-w@163.net
SO CHUNG-HUA I HSUEH I CHUAN HSUEH TSA CHIH, (2000 Oct) 17 (5) 348-51. Journal code: 9425197. ISSN: 1003-9406.
CY China
DT Journal; Article; (JOURNAL ARTICLE)
LA Chinese
FS Priority Journals
EM 200011
ED Entered STN: 20010322
Last Updated on STN: 20010322
Entered Medline: 20001116
AB OBJECTIVE: To determine the distribution of dopamine D2, D3, and D5 receptor(DRD2, DRD3, DRD5) gene polymorphisms in Guangzhou Hans. METHODS: A total of 141 healthy Guangzhou Hans were studied by the use of polymerase chain reaction-restriction fragment length polymorphism (PCR-**RFLP**) and PCR-amplification of specific allele (**PASA**) techniques. Current results were compared with the data on other ethnic groups. RESULTS: Within the 141 individuals tested, the frequencies of the A1(Taq I-) and A2(Taq I+) alleles of the TaqI A mutation site in the 3'non-coding region of the DRD2 gene were found to be 48% and 52%, respectively. The observed frequencies of the A1A1, A1A2 and A2A2 genotypes were found to be 17%, 52% and 31%, respectively, which met Hardy-Weinberg equilibrium. The frequencies of the A1 and A2 alleles of the Bal I mutation site in the exon 1 of DRD3 gene were found to be 73% and 27%, respectively. The observed frequencies of the A1A1, A1A2 and A2A2 genotypes were found to be 53%, 39% and 8%, respectively, which met Hardy-Weinberg equilibrium. The frequencies of the 1 and 2 alleles of the Msp I mutation site in the intron 5 of DRD3 gene were found to be 62.5% and 37.5%, respectively. The observed frequencies of the 1-1, 1-2 and 2-2 genotypes were found to be 35%, 55% and 10%, respectively, which met Hardy-Weinberg equilibrium. No linkage disequilibrium was observed between the Bal I and Msp I polymorphism in Guangzhou Hans(chi(2)=0165, P>0.05). The frequencies of the T and C alleles of the T978C mutation site of DRD5 gene were found to be 51% and 49%, respectively. The observed frequencies of the T/T, T/C and C/C genotypes were found to be 23.6%, 54.6% and 21.8%, respectively, which met Hardy-Weinberg equilibrium. CONCLUSION: The polymorphisms of DRD2, DRD3, DRD5 gene in Guangzhou Hans were high and different from those in other populations.

L7 ANSWER 2 OF 10 CAPLUS COPYRIGHT 2002 ACS
AN 2001:808902 CAPLUS
DN 136:396521
TI **Double ARMS**-PCR analysis and confirmation of the ryanodine receptor gene associated with malignant hyperthermia in swine by a single set of primers
AU Changklungdee, Suwan; Markvichitr, Kanchana; Kanto, Uthai; Vajrabukka, Chanvit; Wajjwalku, Worawidh; Engkagul, Arunee
CS Department of Animal Science, Kasetsart University, Bangkok, 10900,

Thailand
SO Thai Journal of Agricultural Science (2000), 33(3-4), 89-97
CODEN: TJASBN; ISSN: 0049-3589
PB Agricultural Science Society of Thailand
DT Journal
LA English
AB An important advance in DNA amplification technol. for detection of the porcine C1,843 to T mutation of Hal/MH gene is the development of allele-specific PCR or the amplification refractory mutation system (ARMS). The **double ARMS** approach involves using two allele-specific ARMS primers in conjunction with two common external primers simultaneously and Hal genotype detn. can be achieved directly in one step of a single PCR. This method removes the requirement of the current PCR-based test for restriction enzyme digestion and of the single ARMS-PCR. The different Hal genotypes derived from **double ARMS**-PCR can be confirmed its accuracy by sepg. a primer set of **double ARMS** into two subsets of single ARMS for allele-specific reaction (N/n) or into a single subset primers for **RFLP** anal. (NN/Nn/nn). The **double ARMS** approach can be applied to other point mutations in various genomic DNA and it is a method for simply, accurately, reliably, and cost effectively detg. allelic variants.

L7 ANSWER 3 OF 10 CAPLUS COPYRIGHT 2002 ACS
AN 1999:818913 CAPLUS
DN 132:74520
TI Genes for GABAA receptors of insects and their use in screening for novel pesticides and drugs acting of the receptor
IN Ffrench-Constant, Richard H.; Jackson, Meyer B.
PA Ophidian Pharmaceuticals, Inc., USA
SO U.S., 51 pp., Cont.-in-part of U.S. Ser. No. 770,881, abandoned.
CODEN: USXXAM
DT Patent
LA English
FAN.CNT 2

	PATENT NO.	KIND	DATE	APPLICATION NO.	DATE
PI	US 6008046	A	19991228	US 1993-72064	19930602
PRAI	US 1991-770881		19911004		

AB Genes for GABAA receptors of insects, and mutant alleles conferring resistance to cyclodiene insecticides are cloned and characterized for use in methods of screening for novel pesticides and drugs acting on the receptor. Screening methods are disclosed employing cultured cell lines (such as CHO-Pro-3) transfected with plasmid vectors expressing normal and mutant receptors to screen for novel pesticides in large nos. of potentially useful compds. A GABA (.gamma.-aminobutyric acid) type A receptor/chloride ion-channel gene was isolated from Drosophila (D.) melanogaster in search of genes responsible to cyclodiene type pesticide dieldrin. The resistant allele RdlMD-RR contained a point mutation (G.fwdarw.T at nt 995) resulting in restriction enzyme sites polymorphism and Ala.fwdarw.Ser change of the protein. This Ala residue is located at the second membrane spanning region of the receptor and involved to line the Cl- ion channel pore. This mutation is responsible for the resistance phenotype shown by blind testing a larger set of strain using PCR and subsequent digestion with a diagnostic restriction endonuclease and insecticide bioassay. The resistance mechanism was also confirmed by functional study of wild type and mutant cDNA of GABAA locus in the Xenopus oocyte system. A PCR amplification of specific allele (**PASA**) technique was developed using primers contg. the specific single mutated nucleotide at the 3' end for insect genotyping to check its status (susceptible or resistant) to pesticides. **PASA** was successfully applied to D. melanogaster, D. simulans, and three other pest

insects including cockroaches (Perplaneta americana), houseflies (Musca domestica) and red flour beetle (Tribolium castaneum). **PASA** was also applicable to harmful insects since Rdl homolog in mosquito strain A. aegyti possessed analogous Ala.fwdarw.Ser mutation (resulted from G.fwdarw.T at nt 885) which is also responsible for pesticide resistance.
RE.CNT 27 THERE ARE 27 CITED REFERENCES AVAILABLE FOR THIS RECORD
ALL CITATIONS AVAILABLE IN THE RE FORMAT

L7 ANSWER 4 OF 10 CAPLUS COPYRIGHT 2002 ACS
AN 1999:185626 CAPLUS
DN 130:307228
TI Microchimerism analysis using polymerase chain reaction assays that selectively amplify donor DNA
AU Sahota, A.; Yang, M.; McDaniel, H. B.; Hall, M.; Sidner, R. A.; Jindal, R. M.
CS Departments of Medical, Indiana University School of Medicine, Indianapolis, IN, 46202, USA
SO Transplantation Proceedings (1999), 31(1/2), 800-801
CODEN: TRPPA8; ISSN: 0041-1345
PB Elsevier Science Inc.
DT Journal
LA English
AB The presence of donor-type cells in the circulation of organ transplant recipients (microchimerism) may be an important factor in the induction and/or maintenance of allograft tolerance. In this study, the authors evaluated a double amplification refractory mutation system (ARMS-PCR) that has been used previously for the detection of chimerism following bone marrow transplantation. The ARMS-PCR assay is based on polymorphisms in a 3.1-kb region upstream of the .delta. globin gene. Individuals can have two types of sequences in this region, termed R and T, based on digestion of PCR-amplified DNA with the restriction enzyme RsaI or TaqI. RR individuals have an RsaI site at postion 453 and lack a TaqI site at position 1162 on both chromosomes, whereas the reverse is true for TT individuals. RT individuals (heterozygotes) have one chromosome of each type. For microchimerism anal., the genotypes of the donor and recipient at the polymorphic TaqI (or RsaI) positions are first detd. and posttransplant samples from the informative cases are then analyzed using the **double ARMS** assay. Eight of the 48 transplant cases examd. were informative for this assay and, in mixing expts., the authors could detect the equiv. of 0.0001% donor DNA in a total of 100 ng of donor and recipient DNA after 25 cycles of second-round PCR. The amplification was highly specific, since the PCR product was obtained from DNA of the appropriate type only. Using this assay the authors have demonstrated the presence of donor-type DNA in several posttransplant samples from 3 of the informative cases analyzed to date.
RE.CNT 10 THERE ARE 10 CITED REFERENCES AVAILABLE FOR THIS RECORD
ALL CITATIONS AVAILABLE IN THE RE FORMAT

L7 ANSWER 5 OF 10 CAPLUS COPYRIGHT 2002 ACS
AN 1999:80861 CAPLUS
DN 131:307603
TI Genetic polymorphisms in the leptin gene and their association with fatness in four pig breeds
AU Jiang, Zhi-Hua; Gibson, John P.
CS Department of Animal and Poultry Science, Centre for Genetic Improvement of Livestock, University of Guelph, Guelph, ON, N1G 2W1, Can.
SO Mammalian Genome (1999), 10(2), 191-193
CODEN: MAMGEC; ISSN: 0938-8990
PB Springer-Verlag New York Inc.
DT Journal
LA English
AB The leptin gene (OB) of Erhualian, Duroc, and Large White pigs were

sequenced and 4 polymorphisms were discovered. The first 2 (C/T at 867 and A/G at 1112) occurred in introns and the latter 2 (C/T at 3469 and G/T at 3714) in coding regions, but they were silent. The last 3 mutations change the recognition site for enzyme TaqI, HinfI, and PstI; but the PstI site is close to another PstI site and the polymorphism is not easy to sep. on agarose gel. Bi-**PASA** assays were used to assay polymorphisms at sites 867 and 3714; PCR-**RFLP** assays were used for positions 1112 and 3469. From polymorphism anal. of leptin genes in 5 pig breeds, there is a possible assocn. between the polymorphism at 3469 and fatness in pigs.

RE.CNT 17 THERE ARE 17 CITED REFERENCES AVAILABLE FOR THIS RECORD
ALL CITATIONS AVAILABLE IN THE RE FORMAT

L7 ANSWER 6 OF 10 CAPLUS COPYRIGHT 2002 ACS
AN 1997:278258 CAPLUS
DN 126:272956
TI K-ras mutations are found in DNA extracted from the plasma of patients with colorectal cancer
AU Anker, Philippe; Lefort, Francois; Vasioukhin, Valeri; Lyautey, Jacqueline; Lederrey, Christine; Chen, Xu Qi; Stroun, Maurice; Mulcahy, Hugh E.; Farthing, Michael J. G.
CS Departement de Biochimie et de Physiologie Vegetale, Universite de Geneve, Geneva, Switz.
SO Gastroenterology (1997), 112(4), 1114-1120
CODEN: GASTAB; ISSN: 0016-5085
PB Saunders
DT Journal
LA English
AB Circulating DNA can be isolated from the plasma of healthy subjects and from patients with cancer. The aim of this study was to detect K-ras mutations in DNA extd. from the plasma of patients with colorectal cancer. Tumor and plasma DNA were extd. from 14 patients with colorectal cancer (stages A-D), and K-ras alterations were detected using a polymerase chain reaction assay that uses sequence-specific primers to amplify mutant DNA. These results were confirmed with another polymerase chain reaction assay that creates an enzyme restriction site in the absence of a K-ras mutation followed by direct sequencing and addnl. cloning techniques. Seven patients (50%) had a codon 12 K-ras mutation within their primary tumor, and identical mutations were found in the plasma DNA of 6 patients (86%). Mutant DNA was not detected in the plasma specimens of 7 patients whose tumors tested neg. for K-ras alterations or in healthy control subjects. Similar results were obtained using all three mol. biol. techniques. K-ras abnormalities can be detected in circulating DNA extd. from the plasma specimens of patients with colorectal cancer. If these results are confirmed in larger studies, genetic anal. of plasma DNA may have clin. applications in the future.

L7 ANSWER 7 OF 10 MEDLINE DUPLICATE 2
AN 97208461 MEDLINE
DN 97208461 PubMed ID: 9120999
TI Genetic analyses of the ABO blood groups and application of the clinical laboratories.
AU Hosoi E
CS Department of Medical Technology, School of Medical Sciences, University of Tokushima.
SO RINSHO BYORI. JAPANESE JOURNAL OF CLINICAL PATHOLOGY, (1997 Feb) 45 (2) 148-56.
Journal code: 2984781R. ISSN: 0047-1860.
CY Japan
DT Journal; Article; (JOURNAL ARTICLE)
LA Japanese
FS Priority Journals

EM 199704
ED Entered STN: 19970506
Last Updated on STN: 19970506
Entered Medline: 19970422
AB Gene technology using polymerase chain reaction (PCR) has markedly advanced in recent year and has been introduced in clinical laboratories. In this paper, the genotypes of genomic DNAs of subjects with cisAB blood group were analysed using three methods, polymerase chain reaction (PCR)-restriction fragment length polymorphism (**RFLP**), and the PCR-direct sequencing method, and directly determined using the polymerase chain reaction (PCR) amplification of specific alleles (**PASA**)-method. The differences among the methods were as follows, PCR-**RFLP** and PCR-direct sequencing method require 2-step procedures, and are complicated for clinical laboratories. The **PASA** method is based on the fact that PCR amplification occurs only when the 3' endbase of the primer is matched to sites of the nucleotide substitution of ABO allelic cDNA. Three of five regions of allelic DNAs were co-amplified in a single PCR (multiplex-PCR) in this study. ABO and cisAB blood group genotypes were directly determined, based on the molecular size of allele-specific amplification products. The **PASA** method requires only about 4 hours from starting PCR to results, making it rapid, simple and useful for detecting the genotype of ABO and cisAB blood groups in comparison with PCR-**RFLP** and the direct sequencing methods and will allow this procedure to be very versatile and widely used throughout the research and clinical diagnostic communities. The analyses of the nucleotide sequence at nucleotides No. 261, 526, 703, 796 and 803 in 3 major subjects in the cisAB blood group (cisA2B3, cisA1B3 and cisA2B) revealed chimeric structures of the A allele and B allele on the same gene.

L7 ANSWER 8 OF 10 MEDLINE DUPLICATE 3
AN 94324964 MEDLINE
DN 94324964 PubMed ID: 7914079
TI Mismatch PCR **RFLP** detection of DRD2 Ser311Cys polymorphism and schizophrenia.
AU Hattori M; Nanko S; Dai X Y; Fukuda R; Kazamatsuri H
CS Department of Psychiatry, Teikyo University School of Medicine, Tokyo, Japan.
SO BIOCHEMICAL AND BIOPHYSICAL RESEARCH COMMUNICATIONS, (1994 Jul 29) 202 (2) 757-63.
Journal code: 0372516. ISSN: 0006-291X.
CY United States
DT Journal; Article; (JOURNAL ARTICLE)
LA English
FS Priority Journals
EM 199409
ED Entered STN: 19940909
Last Updated on STN: 20000303
Entered Medline: 19940901
AB We tested 100 schizophrenics and 100 controls to find association at the Serine311 Cysteine polymorphism of dopamine D2 receptor in Japanese population. There were no significant differences between the two groups. One homozygote for Cys311 was confirmed in the controls by mismatch-polymerase chain reaction and restriction fragment length of polymorphism (PCR-**RFLP**) analysis. None with Cys was detected among 61 individuals of nine multiply affected families with schizophrenia. Our data thus provide strong evidence against an etiological association between schizophrenia and the Ser311Cys variant. The polymerase chain reaction amplification of specific alleles (**PASA**) analysis was compared with the mismatch PCR-**RFLP** typing, revealing that the latter is more reliable than the former.

L7 ANSWER 9 OF 10 MEDLINE DUPLICATE 4
AN 93138631 MEDLINE
DN 93138631 PubMed ID: 1362561
TI Detection of a new polymorphism of the human prothrombin (F2) gene by combination of **PASA** and mutated primer-mediated PCR-**RFLP**
.
AU Iwahana H; Mizusawa N; Yoshimoto K; Itakura M
CS Otsuka Department of Clinical and Molecular Nutrition, School of Medicine, University of Tokushima, Japan.
SO HUMAN GENETICS, (1992 Nov) 90 (3) 325-6.
Journal code: 7613873. ISSN: 0340-6717.
CY GERMANY: Germany, Federal Republic of
DT Journal; Article; (JOURNAL ARTICLE)
LA English
FS Priority Journals
EM 199302
ED Entered STN: 19930312
Last Updated on STN: 19950206
Entered Medline: 19930225
AB A new polymorphism of the human prothrombin (F2) gene was detected by a combination of polymerase chain reaction (PCR) amplification of specific alleles (**PASA**) and mutated primer-mediated PCR restriction fragment length polymorphism (PCR-**RFLP**). The method is simple and useful for detecting polymorphisms and mutations. The new polymorphism of C1 and C2 examined by this method is highly heterozygous and serves as a good human DNA marker.

L7 ANSWER 10 OF 10 BIOSIS COPYRIGHT 2002 BIOLOGICAL ABSTRACTS INC.
AN 1992:188488 BIOSIS
DN BA93:99438
TI ANALYSIS OF MILK PROTEIN GENES WITH PCR-**RFLP** AND **PASA**.
AU ROTTMANN O; SCHLEE P
CS LEHRSTUHL TIERZUCHT DER TECHNISCHEN UNIV. MUENCHEN, W-8050 FREISING-WEIHENSTEPHAN.
SO ARCH TIERZ, (1992) 35 (1-2), 65-76.
CODEN: ARTZAJ. ISSN: 0003-9438.
FS BA; OLD
LA German
AB The genes coding for bovine milk proteins are highly polymorphic. Some combinations of genes and alleles influence milk performance traits. As the alleles are point mutations on DNA level, they can be diagnosed independently of age and sex. The following loci have been analyzed: alpha S1-, beta- and Kappa-casein and beta lactoglobulin. The method can be applied to distinguish all point mutations in an animal's genome as far as the nucleotide sequence is known.

=> FIL STNGUIDE

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CA SUBSCRIBER PRICE	-3.72	-3.72

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FILE CONTAINS CURRENT INFORMATION.

LAST RELOADED: Aug 30, 2002 (20020830/UP).

=> file medline biosis caplus

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FULL ESTIMATED COST	0.18	38.42

DISCOUNT AMOUNTS (FOR QUALIFYING ACCOUNTS)	SINCE FILE ENTRY	TOTAL SESSION
CA SUBSCRIBER PRICE	0.00	-3.72

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=> s haplotyp? (9a) allel? (9a) (stoffel)
L8 1 HAPLOTYP? (9A) ALLEL? (9A) (STOFFEL)

=> d bib ab

L8 ANSWER 1 OF 1 CAPLUS COPYRIGHT 2002 ACS
AN 2002:104711 CAPLUS
DN 136:146124
TI A PCR-based multiplex assay for determining haplotype
IN Affourtit, Jason Patrick; Seymour, Albert Barnes
PA Pfizer Products Inc., USA
SO Eur. Pat. Appl., 11 pp.
CODEN: EPXXDW
DT Patent
LA English
FAN.CNT 1

	PATENT NO.	KIND	DATE	APPLICATION NO.	DATE
PI	EP 1178119	A2	20020206	EP 2001-306444	20010727
	EP 1178119	A3	20020828		
	R: AT, BE, CH, DE, DK, ES, FR, GB, GR, IT, LI, LU, NL, SE, MC, PT, IE, SI, LT, LV, FI, RO				
	US 2002025530	A1	20020228	US 2001-918203	20010730
PRAI	US 2000-221756P	P	20000731		

AB The invention provides a method for detg. haplotype in a mulriplex version of allele-specific PCR that is conducted in a single reaction tube. The haplotype is the cis arrangement of alleles for two or more polymorphic markers that are located on a single chromosome. The methods for detg. haplotype in a template DNA sequence contg. a first and a second polymorphic marker. The method involves the following steps: (1) comprising combining in a single tube a template DNA sequence, forward primers that are allele-specific for the first polymorphic marker, reverse primers that are allele-specific for a second polymorphic marker, and a Stoffel fragment DNA polymerase; (2) conducting PCR amplifications to produce an amplification product; and (3) analyzing the amplification product to identify which pair of said forward and reverse primers generated said amplification product. The haplotype is detd. by the identification of the forward and reverse primer pair.